1q41q42 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 1q41q42 microdeletion syndrome. ORPHA:250999

1q41q42 microdeletion syndrome is a chromosomal anomaly characterized by a severe developmental delay and/or intellectual disability, typical facial dysmorphic features, brain anomalies, seizures, cleft palate, clubfeet, nail hypoplasia and congenital heart disease.